Galactosemia

Case Presentation

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General Overview

- Inability to break down and absorb galactose
- 1 out of 60,000 Caucasian births
- Galactose makes up half of lactose
- Inherited disease
- There is no definite cure
- Three forms of the disease
  - Galactokinase Deficiency
  - Galactose Epimerase Deficiency
Symptoms and Complications

- Convulsions
- Poor weight gain
- Vomiting
- E. coli sepsis
- Cataracts
- Mental Retardation
- Liver Damage (Cirrhosis)
- Death
Classical Treatment of Galactosemia

Classical Diagnostic Techniques
- Enlarged liver
- Jaundice
- Cataracts (10% of cases)
- Failure to thrive
- Urine tests that yield excessive amounts of ammonia
- Sepsis following aforementioned symptoms indicates high likelihood of Galactosemia

Classical Treatment Techniques
- Dietary Intervention
- Strict Avoidance of All Dairy Products
- Calcium Supplementation
- There is no cure for Classic Galactosemia
Galactose-1-phosphate uridyltransferase enzyme (GALT)

Galactose Metabolism, the Leloir Pathway

Galactose metabolism is important for energy production, glycogen stores, galactosylation of glycolipids and glycoproteins.

Galactose Metabolism, GALT Deficiency

Galactose metabolism is important for energy production, glycogen stores, galactosylation of glycolipids and glycoproteins.
Genetic Information: Autosomal Recessive

- **Homozygotes** for the Classic Galactosemia allele (G/G) exhibit 5% of typical GALT enzyme activity.

- **Heterozygotes** for the Classic Galactosemia allele (G/g) exhibit 50% of standard GALT enzyme activity.
Genetic Diagnostic Tools

- Elimination in breath of less than 5% of $^{13}\text{C}$-galactose as $^{13}\text{CO}_2$ two hours after administration of $^{13}\text{C}$-D galactose can diagnose disease.


- Prenatal and neonatal blood work to measure concentration of GALT enzyme. Less than 5 units of GALT per gram of Hemoglobin indicates Galactosemia.
Novel Therapies

While screening techniques continue to be improved upon due to the genetic understanding of the disease, no viable therapies have been developed to cure Galactosemia.
Citations